

CP 4.1 Primary Congenital Hypothyroidism (PCH) - Follow-up of Positives

Cut-off: The screening test for primary congenital hypothyroidism measures Thyroid Stimulating Hormone (TSH). Newborns are considered screen positive with a TSH > 29 mIU/L.

Policy/Confirmatory Testing:

The NBS Program strongly recommends that PCPs refer babies with positive PCH screens to a pediatric endocrinologist/CCS-approved endocrine center for follow up, or at the very least the PCPs should consult with an endocrine specialist.

The CA NBS Program does not have a contract laboratory to perform confirmatory studies for endocrine positives. All confirmatory testing for primary congenital hypothyroidism (serum thyroid studies) is to be arranged for and ordered by either the primary care physician or a pediatric endocrinologist and conducted by an accredited laboratory. The cost of the testing is not covered by the Program.

The Program follows the recommendations of the American Academy of Pediatrics¹:

1. For newborns with initial TSH values between 29 and 40 mIU/L, treatment should not be initiated until after confirmatory results are available and the diagnosis has been confirmed.
2. For newborns with an initial value of ≥ 40 mIU/L, treatment should be immediately initiated after blood collection for confirmation of diagnosis.
3. However, when an endocrinologist has been consulted, the PCP should follow his/her recommendations.

Panic Value

The NAPS labs are required to repeat the analysis of specimens having an initial positive TSH value that is higher than its analytical range or high standard (which varies with each reagent lot), which is deemed a "Panic Value." Prior to repeating the test, the labs are to report the result to the ASC and enter a Confirmation of Contact in SIS. The ASC is to immediately initiate follow-up by carrying out this protocol. The lab will then report the confirmed result to the ASC as soon as it is available.

Attachments

4.1 A State-Recommended Guidelines for Follow-up of a Positive Newborn Screen for Primary Congenital Hypothyroidism

4.1.B *Why Retest for Primary Congenital Hypothyroidism*

4.1.C California Children's Services-Approved Endocrinology Centers

¹ American Academy of Pediatrics, et al, Update of Newborn Screening and Therapy for Congenital Hypothyroidism, *Pediatrics*, 2006; 117:2290-2303

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PROTOCOL FOR INITIAL POSITIVE RESULTS:

Resp. Person	Action
NAPS Lab	<ul style="list-style-type: none"> • As soon as possible but no later than the end of the same day, calls appropriate ASC when initial TSH result is positive for PCH (including a panic value). • Panic Value: Reports out confirmed result to ASC as soon as it is available. • Enters test result and C of C into SIS.
ASC NBS Coord. Or Program Specialist	<ul style="list-style-type: none"> • As soon as possible, but no later than 48 hours after notification, calls newborn's physician (and the hospital, if infant is still hospitalized) to report results. • Panic Value: Informs PCP/neonatologist that because the TSH value is greater than the highest standard the test is being repeated on the specimen and the result on the mailer will reflect the result of the 2nd test; 2) If confirmed result is negative, informs involved physicians, documents actions in SIS case notes, and discontinues follow-up. • Discusses with PCP/neonatologist the need for confirmatory testing and, if indicated, initiation of treatment as per recommendations noted in Policy above; recommends referral to a CCS-approved Endocrine Center or CCS-paneled Endocrine specialist for diagnosis and treatment (3.19). Assists provider with referral. • Faxes or sends to PCP/neonatologist State-Recommended Guidelines for Follow-up of a Positive Newborn Screen for Primary Congenital Hypothyroidism (Attachment 4.1 A) and list of endocrine centers (4.1 C). • Enters appropriate tracking event(s) and case notes into SIS. • Sends follow-up letter to physician confirming conversation, and encloses educational brochure (Attachment 4.1 B) entitled "<i>Why Retest for Primary Congenital Hypothyroidism</i>". Verifies referral to specific CCS-approved endocrine center, as appropriate. • If baby has been discharged from hospital, also sends parent letter notifying of the need for retesting along with brochure entitled "<i>Why Retest for Primary Congenital Hypothyroidism</i>" (Attachment 4.1 B). • Requests (from primary care provider and/or pediatric endocrinologist) written documentation of diagnosis or rule out of disorder and copy of lab result on which decision is based. When received, files in ASC files.

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	<ul style="list-style-type: none"> Follows case closely until resolved either by confirmed diagnosis with initiated treatment plan (or disorder is ruled out), infant death, noncompliance or lost to follow-up. Documents all attempts at notification, interactions with physicians and parents using tracking events or case notes in SIS. Reports unusual occurrences such as missed cases, anomalous/inconsistent results, lost to follow-up cases, delays in contacting family, delays in analysis or reporting of confirmatory results, etc., of potential significance to the NBSS Nurse Consultant/ASC Contract Liaison. Refers case to Child Protective Services (per protocol) as needed. Resolves case per Case Resolution Protocol (3.20) on case resolution screen in SIS and enters lab test results into SIS Confirmatory Results screen after receiving hard copy of lab report.
Endocrine center staff	<ul style="list-style-type: none"> (Specialist) Contacts PCP to discuss health status of newborn. Determines if immediate visit to SCC is necessary and schedules appointment. At initial visit assists parent/guardian with completion of CCS application. Faxes completed CCS application to local CCS office. Orders confirmatory laboratory tests; provides ASC with test results. Bills CCS for diagnostic services per CCS guidelines. Provides ASC with updates on baby's status through diagnosis, initiation of treatment when indicated. Provides family with educational materials (e.g., <i>Parent's Guide to Primary Congenital Hypothyroidism</i>). If Center has a Vendor Agreement, enters baby information in SIS Endocrine Service Report.